Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platofrm

| Clinical Indication: |
| --- |
| {d.dynamic.clinicalSummary.data} |

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| --- |

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Variant Interpretation & Clinical correlation:

{d.dynamic.FLNC.data}

{d.dynamic.interpretation.data}

{d.dynamic.variantInfo.data.Location}

| Variant Evidence | Gene Impact |
| --- | --- |
| Chromosome: {d.dynamic.variantInfo.data.Location}  Position: {d.position}  {d.dynamic.variantInfo.data.Variation}   | Allele | DP | % | | --- | --- | --- | | {d.dynamic.variantInfo.data.rowData[0][4]} | {d.dynamic.variantInfo.data.rowData[0].} | {d.dynamic.variantInfo.data.rowData[0].%} | |  |  |  |   Genotype:  {d.dynamic.variantInfo.data.rowdata[0][5]}  phred quality score: {d.dynamic.variantInfo.data.phredscore} | RefSeq Gnese 110, NCBI   | Gene:  {d.dynamic.variantInfo.data.rowData[0][2] | Transcript:  {d.dynamic.variantInfo.data.Gene (Transcript)} | | --- | --- | | Effect:  {d.dynamic.variantInfo.data.Classification} | Protein:  {d.dynamic.variantInfo.data.data.rowData[0][4]} | | Exon:  {d.dynamic.variantInfo.data.Variation} | Coding:  {d.dynamic.variantInfo.data.Variation} | |

Based on the evidence, this variant {d.dynamic.variantInfo.data.rowData[0].Variation}

is classified as likely to be {d.dynamic.variantInfo.data.rowData[0].Classification}

variant

OMIM Phenotype

{d.dynamic.variantInfo.data.rowData[0].Disease (OMIM)}

| Case Specific Recommendations: |
| --- |
| {d.dynamic.recommendation.data} |

| Recommendations |
| --- |
| {d.static.recommendation.data} |

| Methodology: |
| --- |
| {d.static.test\_methodology.data} |

| Limitations/Disclaimer: |
| --- |
| {d.static.limitation.data}  {d.static.disclaimer.data} |

Variant Classification as per ACMG guidelines:

| Name | Description |
| --- | --- |
| {d.static.data.rowdata[0][1]} | {d.static.data.rowdata[0][2]} |
| {d.static.data.rowdata[1][1]} | {d.static.data.rowdata[1][2]} |

References:

{d.static.references.data}

Appendix 1: Sample Data and Statistics

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| --- | --- |
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|  |  |

Appendix 2: Coverage Summary

| Mean Depth | Percentage target base pairs covered |
| --- | --- |

| {d.coverageSummary.meanDepth} | {d.coverageSummary.percentageCovered} |  |  |
| --- | --- | --- | --- |
|  |  |  |  |

Appendix 3: Coverage of Analyzed Genes (Percentage of coding region covered)

| Gene | Region Covered | Gene | Region Covered | Gene | Region Covered |
| --- | --- | --- | --- | --- | --- |
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